THE CHILDREN'S ORTHOPEDIC HOSPITAL AND MEDICAL CENTER

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Victor A. McKusick, M.D.
The Moore Clinic
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Dear Victor,

You asked about [redacted], the unusual girl we saw in North Carolina who Charles had seen previously in Texas. To my mind, there are two major possibilities. As you may recall, the parents were quite old at the time she was born and therefore she may represent a new dominant mutation or have been an attempted abortion. With regard to an attempted abortion, there are many features which she has which are reminiscent of Aminopterin induced fetal malformation. Some of those patients have limb deformities. They characteristically have skull defects, craniosynostosis, unusual facies, upswept eyebrows and hair, short stature and mildly decreased IQ's.

The second major possibility is the syndrome Herrmann and Opitz described of craniosynostosis, absent digits, and cranial defects at the Birth Defects meetings in 1969, as well as in the Rocky Mountain Medical Journal, 66:45, 1969. That patient had a very similar facies, cranial defect and limb abnormalities although his feet were more involved than his hands. He was of short stature and mildly retarded.

How did your pictures of her turn out? I would very much appreciate having a copy if they were reasonable.

We recently have seen three or four cases of severely affected juvenile Marfans with significant involvement of the mitral valve. In reviewing your chapter in, Heritable Disorders of Connective Tissue, you seem to suggest that the severely affected children are more likely to have more involvement of the mitral valve. I have two main questions. First, do you have any evidence that there could be two different alleles since most of these young severe cases are new mutations and because of the severe mitral involvment, usually die

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in childhood, rather than going on to reproduce and demonstrate intrafamilial variability. Secondly, does mitral valve involvement seem to protect against aortic aneurysm? Along that same line, with significant mitral involvement would you advocate Propranalol in pre-adolescent children?

Did you happen to see the Newsweek three weeks ago in which there was a picture of a dwarf named Herve Villechaize, p. 44, Newsmakers section? Clinically he looks exactly like [redacted], the little guy that we presented at the Doctor's conference in North Carolina, who had very short hands which were stiff, small facial features and had thyroid nodules and keratoconus of one eye. Do you have influence with movie producers? Apparently he is making a movie in New York and we might be able to find out something about him.

I trust that all is well in Baltimore. We are certainly having a long Indian summer here in God's country.

Sincerely,

Judy

Judith G. Hall, M.D.
Director, Medical Genetics
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and Medicine

JGH/eh